Your Guide to Understanding Genetic Conditions

LAMA2 gene

laminin subunit alpha 2

Normal Function

The *LAMA2* gene provides instructions for making a part (subunit) of certain members of a protein family called laminins. Laminin proteins are made of three different subunits called alpha, beta, and gamma. There are several forms of each subunit, and each form is produced from instructions carried by a different gene. The *LAMA2* gene provides instructions for the alpha-2 subunit. This subunit, together with the beta-1 and gamma-1 subunits, forms the laminin 2 protein, also known as merosin or laminin-211. The alpha-2 subunit, along with the beta-2 and gamma-1 subunits, also forms another laminin called laminin 4, sometimes known as laminin-221.

Laminins are found in an intricate lattice of proteins and other molecules that forms in the spaces between cells (the extracellular matrix). There, the laminins help regulate cell growth, cell movement (motility), and the attachment of cells to one another (adhesion). They are also involved in the formation and organization of basement membranes, which are thin, sheet-like structures within the extracellular matrix that separate and support cells in many tissues. Laminin 2 and laminin 4 play a particularly important role in the muscles used for movement (skeletal muscles). The laminins attach (bind) to other proteins in the extracellular matrix and in the membrane of muscle cells, which helps maintain the stability of muscle fibers.

Health Conditions Related to Genetic Changes

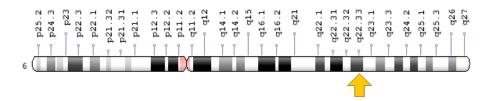
LAMA2-related muscular dystrophy

More than 100 *LAMA2* gene mutations have been identified in individuals with *LAMA2*-related muscular dystrophy, a disorder that causes weakness and wasting (atrophy) of skeletal muscles. This condition generally appears in one of two ways: as a severe, early-onset type or a milder, late-onset form. Most *LAMA2* gene mutations that cause early-onset *LAMA2*-related muscular dystrophy result in the absence of functional laminin alpha-2 subunit. Mutations that cause late-onset *LAMA2*-related muscular dystrophy usually result in a reduction (deficiency) of functional laminin alpha-2 subunit. Deficiency or absence of the laminin alpha-2 subunit results in a corresponding lack of laminin 2 and laminin 4, reducing the strength and stability of muscle tissue and leading to the signs and symptoms of *LAMA2*-related muscular dystrophy.

Chromosomal Location

Cytogenetic Location: 6q22.33, which is the long (q) arm of chromosome 6 at position 22.33

Molecular Location: base pairs 128,883,141 to 129,516,566 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- LAMA2_HUMAN
- laminin-2 subunit alpha
- laminin-4 subunit alpha
- laminin-12 subunit alpha
- laminin M chain
- laminin subunit alpha-2
- laminin subunit alpha-2 isoform a precursor
- laminin subunit alpha-2 isoform b precursor
- laminin, alpha 2
- LAMM
- merosin heavy chain

Additional Information & Resources

GeneReviews

 LAMA2-Related Muscular Dystrophy https://www.ncbi.nlm.nih.gov/books/NBK97333

Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28LAMA2%5BTIAB%5D%29+OR+%28%28laminin+M+chain%5BTIAB%5D%29+OR+%28merosin+heavy+chain%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

 LAMININ, ALPHA-2 http://omim.org/entry/156225

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology http://atlasgeneticsoncology.org/Genes/GC_LAMA2.html
- ClinVar https://www.ncbi.nlm.nih.gov/clinvar?term=LAMA2%5Bgene%5D
- HGNC Gene Family: Laminin subunits http://www.genenames.org/cgi-bin/genefamilies/set/626
- HGNC Gene Symbol Report http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/ hgnc data.php&hgnc id=6482
- NCBI Gene https://www.ncbi.nlm.nih.gov/gene/3908
- UniProt http://www.uniprot.org/uniprot/P24043

Sources for This Summary

- Allamand V, Guicheney P. Merosin-deficient congenital muscular dystrophy, autosomal recessive (MDC1A, MIM#156225, LAMA2 gene coding for alpha2 chain of laminin). Eur J Hum Genet. 2002 Feb;10(2):91-4.
 - Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11938437
- Gavassini BF, Carboni N, Nielsen JE, Danielsen ER, Thomsen C, Svenstrup K, Bello L, Maioli MA, Marrosu G, Ticca AF, Mura M, Marrosu MG, Soraru G, Angelini C, Vissing J, Pegoraro E. Clinical and molecular characterization of limb-girdle muscular dystrophy due to LAMA2 mutations. Muscle Nerve. 2011 Nov;44(5):703-9. doi: 10.1002/mus.22132. Epub 2011 Sep 26. Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21953594

- Geranmayeh F, Clement E, Feng LH, Sewry C, Pagan J, Mein R, Abbs S, Brueton L, Childs AM, Jungbluth H, De Goede CG, Lynch B, Lin JP, Chow G, Sousa Cd, O'Mahony O, Majumdar A, Straub V, Bushby K, Muntoni F. Genotype-phenotype correlation in a large population of muscular dystrophy patients with LAMA2 mutations. Neuromuscul Disord. 2010 Apr;20(4):241-50. doi: 10.1016/j.nmd.2010.02.001. Epub 2010 Mar 6. Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20207543
- Jones KJ, Morgan G, Johnston H, Tobias V, Ouvrier RA, Wilkinson I, North KN. The expanding phenotype of laminin alpha2 chain (merosin) abnormalities: case series and review. J Med Genet. 2001 Oct;38(10):649-57. Review.

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11584042
Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1734735/

- OMIM: LAMININ, ALPHA-2 http://omim.org/entry/156225
- Miyagoe-Suzuki Y, Nakagawa M, Takeda S. Merosin and congenital muscular dystrophy. Microsc Res Tech. 2000 Feb 1-15;48(3-4):181-91. Review.
 Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10679965

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